What's New with RxPups?

This semester the Student Society of Pediatric Advocates (SSPA) at the University of Georgia College of Pharmacy adopted a new name, RxPups - SSPA. RxPups has also added new members, community service projects, and educational opportunities to this year's agenda.

The mission of RxPups is to bring awareness to the proper use of medication therapy in pediatric populations through various service and education-based initiatives. Service activities center around lending our medication-based knowledge to pediatric patients and their parents in our community. Overall, RxPups advocates for the safety and happiness of young patients while learning and having fun along the way. As an organization, we feel that it is our duty as future pharmacist to not only educate ourselves on pediatric pharmacy but also the community around us. The purpose of our newsletter is to educate pharmacy students about pediatric pharmacy.

Over the past year members attended general body meetings and presentations that covered childhood cancer, Kawasaki disease, diabetes management technology, along with several others. Members used these meetings and presentations to gain knowledge about the field of pediatric pharmacy. Throughout the year, members presented on the importance of immunization to students at Pharmtoberfest (UGA College of Pharmacy Health Fair), over-the-counter medication safety to local 1st graders and Girl Scouts at multiple community outreach events. RxPups values giving back to our community. Members volunteered at the Food Bank of Northeast Georgia to pack nutrient rich bags of food for local food-insecure students and their families during the fall and spring semester. The inaugural UGA Miracle College of Pharmacy Fundraising Competition was a success with $2010.00 raised, benefiting Children's Miracle Network. RxPups was also represented at the Pediatric Pharmacy Advocacy Group Spring Conference by President, Nina Del Rosario and Educational Development Coordinator, Gabby Bachner.

The 2018-2019 Executive Board of RxPups is proud to reflect on the accomplishments made over the past year. It is now time to congratulate and welcome the 2019-2020 Executive Board!
Acute Flaccid Myelitis (AFM) is a rare but serious condition that mainly affects young children. AFM and its symptoms are thought to resemble polio and is often referred to as a “polio-like” illness. This condition is characterized by a rapid onset of flaccid weakness of one or more limbs. The patient’s case is further classified as a “confirmed” case if an MRI detects the presence of gray matter lesions of the spinal cord spanning 1 spinal segment, or “probable” if the patient has cerebrospinal fluid (CSF) pleocytosis (> 5 white blood cells/mm³). Other symptoms may include facial drooping and/or weakness, difficulty moving the eyes, drooping of the eyelids, and difficulty swallowing or slurred speech.

The Center for Disease Control (CDC) estimates less than one to two in a million children in the US are diagnosed with AFM annually. Although this is a rare condition, there has been a rise in cases reported since 2014. As of January 4, 2019, the CDC confirmed 193 cases across 39 states within the United States. These confirmed cases are among a total of 349 reports of potential cases received by the CDC. This staggering number vastly differs from the 35 confirmed cases of AFM across 16 states in 2017.

The causes of AFM vary. It’s known that certain viruses are correlated with AFM. Some of those viruses include specific enteroviruses like enterovirus subtypes A71 and D68, poliovirus, and West Nile virus. Since 2014, 90% of patients diagnosed with AFM had a respiratory illness or fever consistent with a viral infection prior to developing AFM. This correlates with the fact that most cases had an onset between August and October, peak time for viruses to circulate in the environment.

If a child experiences any of the symptoms associated with AFM, the CDC urges parents and caregivers to seek medical care immediately. Physicians will begin to take the steps to collect data and images to help reach a diagnosis. This includes assessing the patient’s nervous system via a physical exam and an MRI to look for lesions in the gray matter of the spinal cord. Physicians may also collect samples of the patient’s CSF and stool, as well as conduct a respiratory swab to test for enterovirus, rhinovirus, and poliovirus. Since symptoms of AFM are similar to other neurologic diseases such as transverse myelitis and Guillain-Barre syndrome, testing helps distinguish between these various disease states.

There is no specific treatment or cure for AFM. If diagnosed, a neurologist may refer the patient to a physical or occupational therapist who can recommend different exercises to assist with limb weakness. As mentioned previously, the specific cause of AFM is unknown so there is no prophylactic treatment specifically for AFM. However, since certain viruses are associated with the disease, it’s crucial to get appropriate vaccinations, such as the polio vaccine, to protect yourself and others from developing these viruses which can later progress to AFM. West Nile Virus is also linked to AFM, so techniques to prevent mosquito bites can be used to reduce the chances of developing AFM. This includes the use of mosquito repellent, wearing long shirts and pants to cover the skin, and avoiding being outdoors at dusk and dawn. The best way to prevent getting enterovirus is by washing hands frequently with soap and water.

Pharmacists play a vital role in educating families about the importance of getting vaccinated and practicing good hand hygiene. These two important actions can protect patients from developing this rare condition that is becoming increasingly prevalent. If at any point a patient or caregiver reports the symptoms associated with AFM, such as rapid onset of limb weakness, difficulty moving the eyes, or drooping of the eyes or face, pharmacists should advise the patient to seek medical care immediately.

References:
Neonatal Abstinence Syndrome: Signs, Assessment, and Treatment

Opioid use and abuse has increased to epidemic proportions in the United States over the past decade, leading to an increase in prenatal exposure to narcotics. This exposure causes the development of neonatal abstinence syndrome (NAS). NAS refers to the signs and symptoms that are present in neonates developing from the abrupt cessation of passive transfer of maternal opioids. Clinical presentation of NAS typically occurs within 48 to 72 hours after birth. Symptoms are variable but commonly include tremors, hyperactive moro reflex, high pitched and excessive crying, seizures, diarrhea, vomiting, uncoordinated sucking and swallowing, fever, sweating, and increased respiratory rates. In 2012, incidence of NAS increased to more than 30 per 1,000 hospital live births.

Unfortunately, there is little agreement among researchers and healthcare professionals on the best tools used to assess infants with NAS. However, the Finnegan Neonatal Abstinence Score (FNAS) is the most commonly used scale to screen infants at risk for NAS. FNAS was developed in 1975 and has undergone multiple modifications since. This standardized scoring, shown in Figure 1, allows clinicians to make an assessment, identify and document the infant’s withdrawal symptoms, and initiate appropriate pharmacological therapy if needed.

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Figure 1: http://www.ncpoep.org/guidance-document/neonatal-abstinence-syndrome-overview/neonatal-abstinence-syndrome-nas/
methods recommended include decreasing environmental stimuli by gentle wakening, lowering lighting, and safe swaddling. Mothers fitting certain criteria should be encouraged to breastfeed; breastfeeding decreases the severity of withdrawal symptoms and the need for pharmacological treatment. Mothers who are HIV negative and are on a supervised maintenance program that are compliant to their methadone or buprenorphine therapy and are not using street drugs meet this criteria and should be encouraged to breastfeed. Advantages of breastfeeding a neonate with NAS also include optimal nutrition support and encouragement of mother-infant attachment. Pharmacological therapy for the treatment of NAS is used, but studies of its efficacy are limited. The American Academy of Pediatrics recommends matching drug selection to the type of agent causing the withdrawal. Morphine and methadone are the most prevalent first line agents used to treat NAS; however, there is no evidence supporting which medication is more efficacious. In addition, clonidine, phenobarbital, and buprenorphine can be used alone or as adjunctive therapy to morphine or methadone. Refer to Figure 3 for dosing information.
### Medications to treat neonatal abstinence syndrome

<table>
<thead>
<tr>
<th>Medication</th>
<th>Mechanism of action</th>
<th>Dose</th>
<th>Comments</th>
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<tbody>
<tr>
<td>Morphine</td>
<td>Natural m-receptor agonist, N-methyl-D-aspartate receptor antagonist</td>
<td>If score is a 8 or 2 (less or 2) consecutive evaluations, start at 0.32 mg/kg/day, divided every 4 h, 6 h, or 8 h, orally. If score persists a 8 or 3 (less or 2) consecutive evaluations, increase by 0.16 mg/kg/day (every 4 h, 6 h, or 8 h) to a maximum of 1.0 mg/kg/day. Most tapering protocols decrease dose by 10% of the total daily dose, every 48 h to 72 h, depending on NAS scores. <a href="http://pomch.on.ca/ClinicalPracticaGuidel/NeonatalAbstinenceSyndrome.aspx">http://pomch.on.ca/ClinicalPracticaGuidel/NeonatalAbstinenceSyndrome.aspx</a></td>
<td>Most commonly used as first-line treatment in Canada. Does not contain alcohol. Short half-life (9 h). When NAS scores are stable (&lt; 4) for 48 h to 72 h, consider weaning.</td>
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<tr>
<td>Methadone</td>
<td>Synthetic complete m-receptor agonist, N-methyl-D-aspartate receptor antagonist</td>
<td>0.35–0.71 mg/kg/day every 5 h to 12 h, orally. Increase by 0.03 mg/kg every 48 h. Maximum dose 1 mg/kg/day.</td>
<td>Long half-life (26 h). Used in many countries as a first-line treatment (instead of morphine) when mother is on methadone. Available in Canada but requires special dispensing (prescription authority). Contains 4% alcohol.</td>
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<tr>
<td>Phenoconitol</td>
<td>Gamma-aminobutyric acid (GABA) receptor agonist</td>
<td>May be used in addition to morphine, especially in poly-substance abuse cases. Loading dose: 10 mg/kg, orally, every 12 h for three doses. Maintenance dose: 3 mg/kg/day, orally. Mean by 10% to 20% every day or every two days when symptoms are controlled.</td>
<td>Long half-life (45 h to 100 h). Requires blood level monitoring. May make GI symptoms worse. Sedative effect. Contains 13% alcohol.</td>
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<tr>
<td>Clonidine</td>
<td>Alpha-2-adrenergic receptor agonist</td>
<td>Alternative therapeutic option in combination with morphine. Especially effective when autonomic symptoms of NAS are present. Start at 0.5 mcg/kg/dose every 4 h to 6 h, orally. Mean by 25% of the total daily dose every other day (Q4h to Q5h to Q6h to Q7h to Q8h to Q10h to Q12h to Q15h to Q24h).</td>
<td>Alcohol-free preparation available. Long half-life (44 h to 72 h). Abrupt discontinuation may cause rapid rise in blood pressure (BP) and heart rate (HR). Gradual weaning is therefore recommended.</td>
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<tr>
<td>Buprenorphine</td>
<td>Semi-synthetic partial m-receptor agonist, k-receptor antagonist</td>
<td>4–5 mcg/kg/dose every 4 h or sublingual route. Maximum dose 60 mcg/kg/day.</td>
<td>Half-life (24 h to 50 h). Sublingual administration of a dilution of buprenorphine solution in ethanol and sucrose. Contains 30% alcohol.</td>
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</table>

**Figure 3:** [http://doi.org/10.3389/fped.2018.00033](http://doi.org/10.3389/fped.2018.00033)

All infants who are at risk for developing NAS should be evaluated using a valid scoring instrument such as the FNAS. Non-pharmacological treatments including, encouraging breastfeeding, mother to infant contact, decreasing aggravating environmental factors and pharmacologic measures when indicated are essential to NAS therapy. With increased understanding of NAS, future work creating and modifying NAS assessment and treatment will lead to a comprehensive standardized management of this syndrome that is plaguing the nation.

**References:**


Burosumab Therapy for X-linked Hypophosphatemia
WRITTEN BY NOUR BURJAK, PHARM. D. CANDIDATE 2022

In the United States, approximately 25 to 30 million people are affected by one of 7,000 rare diseases. Patients with rare diseases often have limited to no treatment options. Fortunately, the Office of Orphan Products Development (OOPD) encourages drug companies to develop treatments for these rare diseases through incentives. X-linked hypophosphatemia (XLH) is one of these rare diseases and in April of 2018, the FDA approved a monoclonal antibody therapy, burosumab, to treat and alleviate symptoms of XLH.

XLH is a genetic disorder that affects 1 in 20,000 live births. This disorder is due to mutations in the PHEX gene on the X chromosome. It presents as an increase production of fibroblast growth factor 23 (FGF-23). FGF-23’s main role is to regulate reabsorption of phosphate in the kidneys. However, an overproduction in FGF-23 results in an increased excretion of phosphate through the kidneys and low levels of plasma phosphate. Consequently, this causes skeletal deformities, osteomalacia, and causes bones to soften and weaken, as seen in patients with rickets. The severity of XLH varies from patient to patient. Symptoms are presented similarly among various age groups. For most cases, symptoms first appear within the first 18 months of age. Children may experience abnormal bone development, bone pain, joint pain, a waddling gait, tooth abscesses, and dental pain.

X-linked hypophosphatemia is described as an inheritable, incomplete healing form of rickets which is treated differently. Patients diagnosed with rickets are typically treated with high doses of Vitamin D; whereas patients with XLH are often prescribed a combination of calcitriol and phosphate supplements to alleviate symptoms. Children with XLH begin this therapy at the time XLH is first diagnosed and continue until their bone growth stops. Additionally, growth hormones can be given to improve growth in children with XLH. Adults with XLH remain on calcitriol and phosphate supplements throughout their life to help relieve pain. Other treatment options include corrective surgery to fix bowed or bent legs, skull abnormalities, and dental procedures to treat pain in the teeth and gums.

An open-label, phase two study conducted by Dr. Carpenter and his team evaluated the efficacy and safety of burosumab therapy in children. Burosumab is a recombinant human IgG1 monoclonal antibody therapy that targets and inhibits FGF-23. The team hoped that by inhibiting FGF-23 renal tubular reabsorption of phosphate would increase thereby improving phosphate metabolism. In the study, 52 children between the ages of 5 to 12 years with XLH and active rickets were randomized to receive subcutaneous doses of burosumab every 2 weeks or every 4 weeks. The doses were titrated to achieve low-normal serum phosphorous levels. After 40 to 64 weeks, follow-up measures were assessed.

To measure the efficacy of burosumab, the improvements of rickets in XLH patients were measured and analyzed by the Thacher Rickets Severity Score (RSS). At baseline, XLH patients exhibited deformities such as rickets, bowed legs, and a short stature. By week 40 of treatment with burosumab, the severity of rickets decreased significantly. The mean radiographic Thacher Rickets Severity Total Score decreased from a baseline of 1.9 to 0.8 with 2-week dosing and decreased from a baseline of 1.7 to 1.1 with 4-week dosing. Additionally, the improvements persisted after week 64 (Figure 1).

![Figure 1. Effects of Burosumab on Rickets Scores](https://www.nejm.org/doi/full/10.1056/NEJMoa1714641)
Serum phosphate levels and renal tubular phosphate reabsorption were also analyzed and measured. After the first dose of burosumab, patients showed an increase in mean serum phosphate levels from baseline in both groups. Over half of the patients attained normal levels of serum phosphate by week 6 (3.2 to 6.1 mg per deciliter). At week 40, there was an overall increase in mean serum phosphate levels by 0.75 mg per deciliter (a 34% increase). At week 64, there was an overall increase in mean serum phosphate levels of 0.84 mg per deciliter (a 38% increase). The 2-week dosing maintained stable serum phosphorous levels throughout week 64 while the 4-week dosing showed frequent changes (Figure 2A). Renal tubular phosphate reabsorption increased in both groups. At week 40, there was a total mean increase of 0.98 mg per deciliter (a 51% increase, Figure 2B). At week 64, there was a total mean increase of 1.01 mg per deciliter (a 51% increase, Figure 2B).

![Graph showing changes in serum phosphate levels](https://www.nejm.org/doi/full/10.1056/NEJMoa1714641)

**Figure 2.** Effects of Burosumab on Pharmacodynamic Variables. https://www.nejm.org/doi/full/10.1056/NEJMoa1714641

Patients as young as 1 year of age can be prescribed this medication. The therapy is available as an injectable solution in three different strengths: 10 mg/ml, 20 mg/ml, and 30 mg/ml. For pediatric and adult patients, dosing is based on body weight. For pediatric patients (1–18 years of age), it is recommended to start the regimen at 0.8 mg/kg and administered every 2 weeks. For adult patients (>18 years of age), it is recommended to start the regimen at 1 mg/kg and administered every 4 weeks (Table 1).^5

<table>
<thead>
<tr>
<th>Body Weight (kg)</th>
<th>Starting Dose (mg)</th>
<th>First Dose Increase to (mg)</th>
<th>Second Dose Increase to (mg)</th>
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<tbody>
<tr>
<td>10-14</td>
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<td>105 and greater</td>
<td>90</td>
<td>90</td>
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**Table 1.** Pediatric Dose Schedule for Stepwise Dose Increase. [https://www.crys vita.com/hcp/dosing-and-administration/](https://www.crys vita.com/hcp/dosing-and-administration/)

In August 2018, the FDA approved burosumab (Crysvita) for treatment in XLH patients. Burosumab is the first treatment that directly targets FGF-23, the cause of XLH.^6 Prior to burosumab’s approval, pediatric and adult patients have been managing symptoms of XLH through multiple daily doses of oral phosphate replacement and Vitamin D therapy. Carpenter’s phase II study on burosumab showed improved physical growth, reduced pain, and decreased severity of rickets. Burosumab also improved phosphate homeostasis and overall quality of life in XLH patients. With the data conducted by Dr. Carpenter’s team, along with the recent FDA approval, burosumab is poised to become a vital addition to the treatment and alleviation of symptoms in pediatric and adult patients with XLH.

**References:**

Early Management of X-linked Alport Syndrome

WRITTEN BY: LAUREN ELLARD PHARM.D. CANDIDATE 2021

ALPORT SYNDROME (AS) IS A RARE, HEREDITARY CONDITION THAT IS
CONCOMITANT WITH RENAL, OCULAR, AND COCHLEAR SYMPTOMS.
INDIVIDUALS MAY INHERIT AS THROUGH X-LINKED, AUTOSOMAL RECESSIVE,
OR AUTOSOMAL DOMINANT INHERITANCE. WITH X-LINKED AS COMPRISING
85% OF REPORTED CASES, IT IS CRITICAL TO UNDERSTAND HOW X-LINKED AS
IS APPROPRIATELY MANAGED AND MONITORED AT EARLY AGES, IN HOPES TO
DELAY ONSET OF END-STAGE RENAL DISEASE (ESRD) AND RENAL
REPLACEMENT THERAPY (RRT).

AS IS CHARACTERIZED WITH PERSISTENT GLOMERULAR HEMATURIA,
RETNOPATHY, HEARING LOSS, AND RENAL FAILURE, BY VARYING MUTATIONS
IN THE COL4A5 AND COL4A4 GENES. THE PROGRESSION OF CHRONIC RENAL DISEASE
IN ALL MALES AFFECTED WITH X-LINKED AS BEGINS WITH MICROHEMATURIA,
PROTEINURIA, RENAL INSUFFICIENCY, AND EVENTUALLY ADVANCES TO ESRD.
SENSORINEURAL HEARING LOSS IS USUALLY PRESENT BY LATE CHILDHOOD.
HOWEVER, IN SOME CASES IT IS NOT EVIDENT UNTIL EARLY ADOLESCENCE.
FEMALES THAT INHERIT X-LINKED AS WILL PRESENT DIFFERENTLY THAN
AFFECTED MALES. AFFECTED FEMALES WILL HAVE ASYMMPTOMATIC
HEMATURIA AND A 50% CHANCE TO PASS THE GENETIC MUTATION TO ANY
OF HER OFFSPRING. MALES WITH X-LINKED AS WILL PASS THE MUTATION ON
to ALL OF THEIR DAUGHTERS, BUT NONE OF THEIR SONS.

SINCE AS HAS SYMPTOMS SIMILAR TO OTHER RENAL DISEASES, LIKE THIN
BASEMENT MEMBRANE NEPHROPATHY (TBMN), IT MUST BE CLINICALLY
DISTINGUISHED BETWEEN CONDITIONS PRESENTING WITH PERSISTENT
GLomerular Hematuria. AS Is Highly Likely if the Patient Has
Retinopathy or Lenticonus. Unlike TBMN, which presents with
Glomerular basement membrane thinning, AS develops with
Glomerular basement membrane lamellation.

IT IS IMPERATIVE TO DiAGNOSE AS EARLY, TO DELAY AND MANAGE THE
ADVANCEMENT OF ESRD IN MALE X-LINKED AS INDIVIDUALS, ESPECIALLY
FOR MUTATIONS WITH EARLY ONSET OF SYMPTOMS THAT NECESSITATE
MUCH MORE AGGRESSIVE TREATMENT. DIAGNOSIS OF AS BEGINS WITH
OPHTHALMIC, AUDILOGIC AND RENAL EVALUATIONS. A RENAL BIOPSY
THROUGH ELECTRON MICROSCOPY AND A FAMILIAL MEDICAL HISTORY
REPORT ARE NEEDED FOR ANALYSIS OF X-LINKED AS. IN X-LINKED AS, THE
NONSENSE AND FRAMESHIFT MUTATIONS THAT OCCUR LEAD TO A 50%.
CHANCE OF REACHING ESRD BEFORE THE AGE OF 20. IF THE AS PATIENT
HAS A LARGE FRAMESHIFT, NONSENSE, OR SPlice SITE VARIATION OF
COL4A5, THEY ARE AT A 50% RISK OF DEAFNESS BY THE AGE OF 10 YEARS
OLD. HOWEVER, IF THE AS PATIENT Has A Missense Mutation of
COL4A5, THEN THEY HAVE A 50% CHANCE OF DEAFNESS BY THE AGE OF
20. Studies have also shown that podocyte loss around the
glomerulus leads to ESRD in AS patients by an average age of 22
years.

CURRENTLY, THERE ARE NO SPECIFIC TREATMENTS FOR X-LINKED AS. ALL
MALE INDIVIDUALS WILL EVENTUALLY ACQUIRE ESRD. HOWEVER, THERE ARE
TREATMENT ACTIONS FOR THE HYPERTENSION AND PROTEINURIA THAT OCCUR
WITH THE DISEASE IN ORDER TO DECELERATE THE ADVANCEMENT OF ESRD.

BY REDUCING ONE’S BLOOD PRESSURE, LESS STRESS IS PLACED ON THE
GLomerulus. IF ESRD DOES OCCUR, PATIENTS ARE TREATED WITH DIALYSIS
AND RENAL TRANSPLANTATION. TREATMENT STARTED EARLY WITH AN ACE
INHIBITOR MAY HELP DELAY THE ONSET OF ESRD, WHICH OCCURS IN
APPROXIMATELY 50% OF PATIENTS BEFORE THE AGE OF 20. ACE
INHIBITORS HAVE BEEN SHOWN TO REDUCE PROTEINURIA IN X-LINKED AS
PATIENTS WHICH MAY HELP POSTPONE RENAL FAILURE.

IN EUROPE, A STUDY WAS INITIATED ON SEVERAL GENERATIONAL FAMILIES
WITH AS CATEGORIZED BY RENAL FUNCTION AND THE INTRODUCTION OF
TREATMENT. A TOTAL OF 283 PATIENTS WERE FOLLOWED FOR OVER 20 YEARS
TO EVALUATE THE EFFECT OF ACE INHIBITOR TREATMENT IN DELAYING THE
PROGRESSION OF ESRD IN COMPARISON TO A CONTROL GROUP WITH NO
TREATMENT. THOUGH OTHERS MAY BE CHOSEN, THE MOST COMMONLY
USED ACE INHIBITORS IN THE STUDY CONSISTED OF RAMIPRIL AND
ENALAPRIL. IN UNTREATED PATIENTS, THE MEDIAN AGE OF ESRD WAS 22
YEARS OLD. HOWEVER, PATIENTS TREATED BY THE AGE OF 13 WITH AN ACE
INHIBITOR AND A MEAN DURATION OF 5.8 YEARS SHOWED A DELAY IN THE
AGE OF ESRD BY A RANGE OF 18-40 YEARS. THIS RANGE WAS COMPARED TO
THE AGE OF 22 YEARS FOR THOSE UNTREATED (P<0.0001). THIS STUDY
ALSO DEMONSTRATED THAT CHILDREN TREATED BY THE AGE OF 8 YEARS
WITH A TOTAL THERAPY DURATION OF 4 YEARS HAVE YET TO EVEN REACH CKD
STAGE 3 OR GREATER (P<0.001). TO DETERMINE THAT THIS STUDY WAS
CONDUCTED IN A NONBIASED DESIGN, THE SAME STUDY METHODS WERE
PERFORMED ON 15 DIFFERENT GROUPS OF SIBLINGS THAT PRESENTED WITH
IDENTICAL MUTATIONS AND ENVIRONMENTS. THE STUDY SHOWED THAT
DIAGNOSES IN OLDER SIBLINGS ALLOWED FOR EARLY DETECTION AND
TREATMENT IN THEIR YOUNGER SIBLINGS, PERMITTING THE DELAY IN RRT
UP TO 40 YEARS OF AGE (P<0.001). ALTHOUGH ACE INHIBITORS ARE NOT
SPECIFIED FOR TREATMENT OF X-LINKED AS, THESE STUDY FINDINGS HELP
ENCOURAGE EARLY TREATMENT AND THE NEED FOR A TIMELY DIAGNOSIS AT A
YOUNG AGE.
In children diagnosed with X-linked AS, routine treatment for hypertension, sensorineural hearing loss, and cataract removal should be implemented. The children should minimize exposure to loud noises and continue audiologic evaluations yearly, beginning at the age of 6. Affected children’s hearing will continue to decline into adulthood but can be improved through the use of hearing aids. The response to pharmacologic treatments will vary with all X-linked AS patients, but it is important to begin therapy early before the onset of microalbuminuria. Though clinical management may help reduce symptoms and delay onset, no treatment is likely to be completely effective.

Concerns with regards to testing for AS at an early age for patients with a family history of it are far outweighed by the potential to delay ESRD. Affected female individuals should consider reproductive options, such as prenatal and preimplantation genetic diagnosis prior to any pregnancy. Because ACE inhibitors reduce proteinuria in children with X-linked AS, treatment before the onset of proteinuria, in children below the age of 13, with the genetic alterations of COL4A5 or COL4A3 and COL4A4 and with a family history of the disease may suspend the onset of ESRD. Preventing ESRD from occurring in youth will enhance life expectancy in persons with X-linked ESRD.

References:


